

A better future for baby: Stemming the tide of fetal alcohol syndrome

These strategies can help you recognize an infant impaired with FAS. Screening tools like TWEAK can help you spot mothers at risk.

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PRACTICE RECOMMENDATIONS

› Screen all females of child-bearing age for alcohol use with standardized screening instruments such as TWEAK, T-ACE, or AUDIT. **(A)**

› Use brief, in-office interventions to decrease alcohol consumption and increase effective contraception in sexually active women. Involve a supportive adult of the patient's choosing where possible. **(A)**

› Advise pregnant patients that complete abstinence from alcohol is safest. **(B)**

› Refer a child who meets FAS referral criteria for multidisciplinary evaluations as early as possible. **(A)**

Strength of recommendation (SOR)

- (A)** Good-quality patient-oriented evidence
- (B)** Inconsistent or limited-quality patient-oriented evidence
- (C)** Consensus, usual practice, opinion, disease-oriented evidence, case series

Family physicians who care for infants and young children are often asked to diagnose growth lags and failure to meet parents' expectations for reaching developmental milestones: Why isn't my child gaining weight? Why isn't he talking? The other kids in the day-care center ride tricycles—why can't he? Will he catch up? My mother thinks he's funny looking. Do you?

Simple reassurance is all that most of these worried families need. But for families with children whose growth or developmental milestones are sufficiently outside the usual parameters, more than reassurance is called for. As you consider whether the lags that worry parents are signs of a serious disability, it's important to make a place for fetal alcohol syndrome (FAS) in your differential.

FAS, a congenital disorder caused by alcohol exposure during pregnancy, is characterized by growth deficiency before and after birth, distinctive facial features, and central nervous system (CNS) dysfunctions. The cognitive and developmental effects of FAS persist throughout life and are severe enough to limit employment and independent living.^{1,2}

■ **A spectrum of severity.** FAS is the most severe expression of prenatal alcohol exposure. The term fetal alcohol spectrum disorders (FASD) is a nondiagnostic umbrella term that includes FAS as well as ARND (alcohol-related neuro-behavioral disorder) and ARBD (alcohol-related birth defects). Children with ARND and ARBD fail to meet the full FAS diagnostic criteria but still exhibit the negative effects of gestational alcohol exposure. Centers for Disease Control and Prevention (CDC) diagnostic criteria for FAS are summarized in **TABLE 1**. Studies by the CDC have reported FAS prevalence rates from 0.2 to 1.5 cases per 1000 live births, with a higher prevalence among minority (Native American and African American) and impoverished groups.¹

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TABLE 1

Diagnostic criteria for fetal alcohol syndrome

Facial dysmorphism <ul style="list-style-type: none"> • Shortened palpebral fissures • Smooth philtrum • Thin vermilion border of upper lip 	≤10th percentile for age and racial norms Score of 4 or 5 on lip-philtrum guide* Score of 4 or 5 on lip-philtrum guide*
Growth problems	<10th percentile for age, sex, gestational age, racial norms in height or weight, prenatally or postnatally
CNS abnormalities	Any structural abnormality (head circumference <10% of age norm or clinically significant brain abnormalities observable through imaging); neurological abnormality not due to postnatal insult or fever; or functional abnormality demonstrated by cognitive performance less than expected for age, schooling, or family circumstances. An individual could meet the CNS abnormality criteria for a FAS diagnosis through a structural abnormality, yet not demonstrate detectable functional deficits.

CNS, central nervous system.

*The lip-philtrum guide is available at <http://depts.washington.edu/fasdpn/htmls/lip-philtrum-guides.htm>.

Source: Centers for Disease Control and Prevention. *Fetal Alcohol Syndrome: Guidelines for Referral and Diagnosis*. 2005. Available at www.cdc.gov/ncbddd/fasd/documents/FAS_guidelines_accessible.pdf. Accessed April 10, 2010.

Correctly diagnosing a child with FAS before age 6 can decrease the odds that he or she will suffer severe secondary disabilities in adolescence and adulthood.

■ An opportunity—and a challenge. As a family physician, you have a unique opportunity to modify the impact of FAS by recognizing the disorder in infancy or early childhood, actively engineering appropriate referrals, and supporting families in the difficult task of parenting a child with disabilities. Correctly diagnosing a child with FAS before age 6 can have a protective influence, decreasing the odds that he or she will suffer severe secondary disabilities in adolescence and adulthood. You can also help prevent FAS by screening for potentially harmful drinking patterns and helping sexually active female patients decrease alcohol consumption and use contraception successfully.

Providing these supportive and preventive services can be challenging. A recent survey of pediatricians revealed that only 34% felt prepared to manage and coordinate the treatment of children with FASD, and only 13% routinely counseled adolescent patients about the risks of drinking and pregnancy.³ This article will help you surmount the difficulties these tasks present and perform vital functions for alcohol-affected families you may encounter in your practice.

The place to start: Spotting mothers at risk

Recognizing an infant with FAS starts by asking the baby's mother about her pattern of drinking while she was pregnant. Most studies on the effects of gestational exposure to alcohol have emphasized moderate to high levels of exposure. In 1 study, children who were exposed to binge drinking were 1.7 times as likely to have IQ scores in the mentally retarded range and 2.5 times more likely to have clinically significant levels of delinquent behavior.⁴ Binge drinking is defined by the National Institute of Alcohol Abuse and Alcoholism as a pattern of drinking that brings blood alcohol concentration to 0.8% or above, which typically happens in women who consume 4 or more drinks in a period of about 2 hours.⁵

But a pregnant woman doesn't have to be a binge drinker to put her fetus at risk. Even low levels of prenatal alcohol use—as low as 1 drink per week—have been associated with adverse behavioral changes in children, including increased aggressive behaviors documented at school age.^{6,7} The research documenting effects at these low levels has led the American Academy of

Pediatrics (AAP) and the American Congress of Obstetricians and Gynecologists (ACOG) to recommend total abstinence from alcohol throughout pregnancy.^{8,9} Patterns of “at risk” drinking for women include binge drinking or persistent regular use (>7 drinks in 1 week). If a mother provides a history of that level of prenatal exposure, her child should be referred for multidisciplinary evaluation at an FAS center, *even in the absence of the characteristic facial features*.¹

The face of FAS

Without a history of prenatal alcohol exposure, the cardinal features of facial dysmorphism (short palpebral fissures, smooth philtrum, and thin vermilion border) plus deficits in height and weight are the main physical findings of FAS. Evaluating height and weight percentiles is a routine part of well-child care, requiring minimal training. Height or weight or both at or below the 10th percentile, adjusted for age, sex, gestational age, and race or ethnicity, meet part of the CDC diagnostic criteria for FAS, but must be accompanied by at least 1 of the typical facial features associated with FAS to meet *referral* guidelines.¹ The *diagnostic* guidelines are more restrictive, requiring all 3 facial features to meet the threshold for an FAS diagnosis, vs another diagnosis, such as alcohol-related neurodevelopmental disorders (TABLE 1).

■ **Learning to measure faces.** Examining a child for the facial characteristics of FAS requires a set of skills that can be learned in a relatively short time, with moderate interrater reliability when compared with dysmorphologists, according to 1 study.¹⁰

■ **Tools for measuring.** Palpebral fissure length (PFL) can be measured with a clear plastic ruler pressed onto the child’s cheek to determine the distance from the endocanthion to the exocanthion while the child is gazing upward.¹¹ To meet the CDC criteria for FAS, this distance should be at or below the 10th percentile compared to norms. The shape of the lips and the nature of the philtrum are then compared to preestablished comparison photographs available on the University of Washington Lip-Philtrum Guides (see URL that follows).¹² The vermil-

Fetal alcohol syndrome across the lifespan

Changes in the clinical presentation of fetal alcohol syndrome (FAS) occur across the lifespan. Clinicians searching for an explanation of an older child’s difficulties may need to reconsider a possible diagnosis of FAS, even if the facial features are less recognizable.

Preschool children with FAS may display a delay in early language acquisition and the beginnings of attention problems and hyperactivity. School-age children may have difficulty remembering material even when they have gone over it many times. They may lack social perception skills, be hyperactive, and show cognitive delays. Mathematics is often an area of severe delay, and sleep problems can persist.¹

Teens and adults with FAS often have a wide range of secondary disabilities: disrupted schooling, inappropriate sexual behavior, mental health problems, aggression, trouble with the law, confinement in jail or as inpatients in psychiatric or substance abuse treatment facilities, alcohol and drug problems, and failure or inability to live independently at an appropriate age. They may have trouble finding a job and keeping one. More than 90% of patients with FAS exhibit mental health problems, with attention problems appearing in early childhood and persisting, and depression appearing in adolescence and adulthood.¹⁵

lion border and the philtrum must both receive a rank of 4 or 5 to meet FAS criteria.¹¹

Tools available online for physicians include Lip-Philtrum Guides (www.depts.washington.edu/fasdpn/htmls/lip-philtrum-guides.htm) and an instructional video depicting PFL measurement techniques (www.depts.washington.edu/fasdpn/htmls/photo-face.htm), both from the University of Washington FAS Diagnostic and Prevention Network. In addition, a physical evaluation summary form, with reference data for PFL in Caucasian, black, and Hispanic children, is available at <http://www.fas.academicedge.com/documents/phyevaln.pdf>.

■ **The Astley-Clarren criteria.** The 4-digit diagnostic code developed by Astley and Clarren for diagnosis of FAS and employed at some FAS referral centers uses very strict criteria.¹³ Centers using these criteria define “abnormal” as ≥ 2 standard deviations below the mean or its equivalent, ≤ 2.5 th percentile.¹³ This applies to the 3 facial features and CNS dysfunction

> Even low levels of prenatal alcohol use—as little as 1 drink per week—have been associated with adverse behavioral changes in children, including increased aggression.

(low IQ, eg). If all 3 facial features are identified in the Astley-Clarren system as abnormal (PFL \leq 2.5th percentile, lip philtrum 5, vermilion 5), the sensitivity of the facial features is 100% and specificity is 99.8% for a diagnosis of FAS.¹³

The CDC criteria, developed with increased surveillance by providers as a goal, uses relaxed criteria of \leq 10th percentile of PFL and 4 or 5 on the philtrum/vermillion border guide to identify abnormal facial features and $<$ 10th percentile in CNS dysfunctions. Sensitivity and specificity data for those CDC criteria are not available.¹

■ **CNS abnormalities** may be noted early enough to trigger a referral for complete evaluation, but must be present in some degree to confirm a diagnosis of FAS. Abnormalities may include microcephaly with head circumference below the 10th percentile; clinically significant brain abnormalities observable through imaging, especially a small or absent corpus callosum; and functional deficits in any of a multitude of domains. In an infant, these deficits may be expressed in global developmental delays, sleep cycle problems, poor muscle tone, and feeding problems with poor suck and texture aversion.¹

■ **Evaluation may not confirm the diagnosis.** Children referred for more extensive evaluation may or may not be confirmed to have FAS. In 2 demographically similar counties in New York state, only 5% of children initially identified in 1 county (10 of 208) and 13% (53 of 420) of children in the other county were confirmed to have FAS.¹² The FAS diagnosis is complicated and the CNS and growth deficiencies may not be expressed until a later age.¹⁴ (See “Fetal alcohol syndrome across the lifespan”^{1,15} on page 339.)

Providers may feel reluctant to alarm or stigmatize families when they are unsure of the diagnosis, but the long-term benefit of confirming the diagnosis early on may be significant for the child and family. The case on page 341 (Tanya) illustrates the complexity of diagnosing FASD.

■ **The encouraging news for family physicians** is that the odds of escaping adverse life outcomes are increased 2- to 4-fold by receiving a diagnosis of FAS before age 6

and by being raised in a stable environment.¹⁶ Early diagnosis can be protective by helping with eligibility requirements for support services and by opening the door to medical management of FAS-associated conditions such as ADHD and depression. In addition, the diagnosis can alert family physicians to the family’s need for help with ongoing problems with alcohol use. The case on page 341 (Brianna) illustrates the complex secondary problems a teen with FAS may face.

Putting families in touch with resources

Multidisciplinary FAS teams may include physicians (a geneticist or developmental pediatrician), psychologists, speech pathologists, educational specialists, social workers, and occupational therapists. These groups typically have in-depth intake and evaluation processes, including neurodiagnostic studies that help clarify the cognitive and functional domains that are affected.

You can locate the nearest FAS evaluation team and other resources for providers and families on the National and State Resource Directory for the National Organization on Fetal Alcohol Syndrome. Go to www.nofas.org, click on Resources, then on National and State Resources Directory in the box on the left side of the page. There may be a waiting list for evaluation, but under Part C of the Individuals with Disabilities Education Act (IDEA), FAS is considered a “presumptive eligibility” diagnosis. Presumptive diagnoses allow children under age 3 at risk of later developmental delay to be served without meeting particular eligibility criteria.¹ Physicians may refer these children for developmental assessment services and early intervention services while waiting for the more complete FAS evaluation.

■ **After the age of 3**, children and families are referred to preschool programs for children with disabilities that are administered through IDEA Part B, with no “presumptive eligibility” diagnoses. Eligibility for educational services under this program is entirely based on functional criteria.

Your best bet: Prevention

The key to preventing FAS is to find out whether your patient’s drinking patterns and

Making the FAS diagnosis: 3 cases*

TANYA

Possible, though unconfirmed, FAS

This 1-month-old African American girl was admitted to the hospital with stridor, possible cardiorespiratory issues, and failure to thrive. She exhibited microcephaly, poor suck, floppy overall muscle tone, and small palpebral fissure lengths. The respiratory noise was attributed to tracheomalacia, secondary to her poor muscle tone.

An initial magnetic resonance imaging of the brain showed an almost complete absence of the corpus callosum. Obviously at risk for future delays, Tanya was referred for early childhood development intervention. By the age of 5½, she was within her peer group's normal range in fine motor, gross motor, and speech skills. She was removed from her mother's care for neglect and later placed in an extended family adoption.

Her initial evaluation took place before the widespread publication of fetal alcohol syndrome (FAS) criteria, and she was not referred for more specific FAS evaluation, as she would have been had she been seen more recently. Tanya has been lost to long-term follow-up from her initial medical home, but her mother returned to the same practice for a subsequent pregnancy and was screened as a problem drinker.

BRIANNA

Facing severe secondary disabilities

This 16-year-old Caucasian girl entered foster care for the second time because her first placement could not deal with her complex behavioral issues. Her mother committed suicide last year, and her father is in an alcohol rehabilitation program. No pregnancy history is available.

The social service agency supervising Brianna's care decided not to return her to her father's home. She made a suicidal gesture, her behavior is sometimes violent, and she has been diagnosed with attention deficit hyperactivity disorder (ADHD). She is being treated for depression, requiring multiple medications.

Brianna's life has been difficult. Before she was 2 years old, she underwent surgery for a ventricular septal defect. Although her cardiac repair was successful, she remains significantly underweight, <3rd percentile on the growth curve. Because she is an adolescent, her facial features are less distinctive for FAS, but she appears to have a smooth philtrum and thin vermilion. Her learning disabilities are significant enough that she has been held back a grade and requires an individualized education plan.

It may be too late to help provide any assistance to Brianna, who is already experiencing severe secondary disabilities. But if a diagnosis of FAS (or alcohol-related neurobehavioral disorder) can be made, even at this point in her life, provisions may be made within the foster care system for transitional housing and emphasis on life skills training, rather than simply allowing her to "age out" of the system when she turns 18.

CLARICE

Preventing an alcohol-exposed pregnancy

Clarice is an 18-year-old, college-bound woman who came in for a pre-college health maintenance examination. She reported being sexually active and was using effective contraception only intermittently. On the TWEAK questionnaire, she said the most she has had to drink on a single occasion was 6 drinks, noted that she had never passed out and had not had any problems associated with drinking. The only worry she connected with drinking was a fear of gaining weight.

Clarice met the criteria for "at risk" drinking (TABLE 3). She was a candidate for a brief intervention, including information on the hazards of alcohol-exposed pregnancy and on effective contraceptive practices. We scheduled a telephone follow-up and checkup in 6 months.

* Drawn from the author's case files. Names have been changed to protect patient privacy.



The cardinal features of facial dysmorphism—short palpebral fissures, smooth philtrum, and thin vermilion border—plus deficits in height and weight are the main physical findings of fetal alcohol syndrome.

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Teens and adults with fetal alcohol syndrome may exhibit a range of disabilities, including inappropriate sexual behavior, mental health problems, and an inability to live independently.

TABLE 2

TWEAK your patients for alcohol use

Tolerance: How many drinks does it take for you to:	Score
• Feel the first effects of alcohol?	2 points for ≥3 drinks
• Fall asleep or pass out? Or, if you never pass out, what is the largest number of drinks you have on 1 occasion?	2 points for ≥5 drinks
Worry about drinking	
Have your friends or relatives worried about your drinking in the past year?	1 point for Yes
Eye-opener	
Do you sometimes take a drink in the morning when you first get up?	1 point for Yes
Amnesia	
Are there times when you drink and afterwards can't remember what you said or did?	1 point for Yes
(K)Cut down	
Do you sometimes feel the need to cut down on your drinking?	1 point for Yes

Scoring: ≥3 points is considered positive for alcoholism/heavy drinking. Thresholds differ for screening different populations, and a score of ≥2 points should be used as the threshold for identifying female problem drinkers.

Source: Chan AW, et al. *Alcohol Clin Exp Res*. 1993.¹⁹

contraceptive habits put her at risk for an alcohol-exposed pregnancy. Make it routine practice to ask women, in a way that encourages honest reporting, about both of these aspects of their lives. The US Preventive Services Task Force recommends screening and counseling intervention in primary care settings to reduce alcohol misuse in adults, including pregnant women.¹⁷ The case on page 341 (Clarice) illustrates how screening and brief intervention can be used to prevent alcohol-exposed pregnancy.

Screening should include simple quantity and frequency questions developed by the National Institute on Alcohol Abuse to clarify a patient's current drinking patterns. The questions include the numbers of days per week of any drinking, the average number of drinks per day, and the maximum number of drinks consumed in 1 day during the past month. Determining that a woman drinks more than 7 drinks per week has a 29% sensitivity, but a 90% specificity for identifying lifetime risk of alcohol abuse or dependence.¹⁸

Other tools include TWEAK (Tolerance, Worry, Eye-opener, Amnesia, (K)Cut down), T-ACE (Tolerance, Annoyed, Cut down, Eye opener), and AUDIT (Alcohol Use Disorder Identification Test). They are detailed below and available online at Project Cork. Go to www.projectcork.org, and click on "clinical tools."

■ **The 5-item TWEAK tool** (TABLE 2) appears to be the optimal screening questionnaire for identifying women in racially mixed populations with heavy drinking or alcohol abuse and dependence, but a score of 2 points should be the threshold for identifying female problem drinkers.^{19,20} Using the lower cutoff of 2, the sensitivity of TWEAK is in the 87% to 91% range for women, with a specificity of 77% to 90%.¹⁸

■ **The T-ACE tool** is a set of 4 questions, with the question addressing tolerance weighted more heavily than the others. With a score of 2 or more, the sensitivity of T-ACE is 70% to 88%; the specificity is 79% to 85%.¹⁸

■ **The AUDIT tool** is a self-administered screen that consists of a series of 10 questions

TABLE 3

Drinking patterns in women

<p>Abstainer</p> <ul style="list-style-type: none"> • Does not drink alcohol or has <12 drinks per year
<p>Low-risk drinking</p> <ul style="list-style-type: none"> • Has ≤7 drinks per week • Has ≤1 standard drink on a single occasion • Abstains before driving, when pregnant, nursing, or while taking contraindicated medications • Has not suffered any negative consequences from drinking.
<p>At-risk drinking</p> <ul style="list-style-type: none"> • Has >7 standard drinks per week or >3 standard drinks on any 1 occasion • Has any amount of alcohol when pregnant or nursing • Has not experienced any negative consequences from drinking.
<p>Problem drinking</p> <ul style="list-style-type: none"> • Has >7 standard drinks per week or >3 standard drinks on any 1 occasion • Has any amount of alcohol when pregnant or nursing • Has experienced negative medical or social consequences from drinking.* <p>*Negative medical consequences include peptic ulcer disease, gastroesophageal reflux disease (GERD), hypertension, obesity, depression, or liver disease. Negative social consequences include receiving a summons for driving under the influence (DUI), involvement in a motor vehicle accident, relationship problems or divorce, and loss of employment.</p>
<p>Alcohol-dependent drinking: Maladaptive patterns of alcohol use leading to clinically significant impairment manifested by 3 or more of the following, occurring at any time during the same 12-month period:</p> <ul style="list-style-type: none"> • Has experienced tolerance • Has experienced withdrawal • Drinks greater amounts than intended • Has made unsuccessful attempts to cut down or control drinking • Spends significant amounts of time obtaining alcohol, drinking, and recovering from drinking • Has given up important activities, or spends less time on them • Continues to use alcohol despite knowledge of physical or psychological problems caused or worsened by alcohol.

Source: Training program for medical students and other health professionals on fetal alcohol syndrome. Presented by the Midwest Regional Fetal Alcohol Syndrome Training Center; March 18, 2005; St. Louis, Mo.

that are each scored on a scale of 0 to 4. The maximum score is 40. A score of 2 indicates some harmful use of alcohol, but a score of 8 or more has a sensitivity of 59% to 66%, with a specificity of 93% to 97% in women.¹⁸

When your patient is at risk

Brief interventions are recommended for nonpregnant and pregnant women who have exhibited a pattern of at-risk or problem drinking.²¹ TABLE 3 summarizes patterns of drinking in women, from not drinking at all through various degrees of risk to alcohol dependency. Referral to an addiction specialist is recommended for women with alcohol dependence.

■ **Brief interventions** for alcohol abuse can be single-session encounters from 5 to 15 minutes' duration, or multi-contact brief sessions, including possible phone follow-up contacts. Project TrEAT (Trial for Early Alcohol Treatment) provided two 15-minute sessions with the primary care physician (either a family physician or internist in community-based practice) scheduled 1 month apart, with nurse follow-up phone calls 2 weeks after each appointment.²²

Patients received general information regarding adverse effects of alcohol and the prevalence of problem drinking, in addition to tools to help them identify drinking triggers and track their consumption. The patient and physician developed a "drinking agreement" in the

Make it routine practice to ask women about their drinking patterns and contraceptive habits.

Brief interventions for alcohol abuse can take as little as 5 minutes.

form of a prescription. Men and nonpregnant women were included in this study.

Women reduced their alcohol use by 47% and their frequency of binge drinking by 56%, as noted at a 6-month follow-up, with changes well maintained at 12 months. The reductions for female patients were actually slightly higher than for male participants.²²

Chang and colleagues provided a 25-minute single session brief intervention to pregnant women who had screened positive on the T-ACE questionnaire with a score of 2 or more, and were identified as being at risk for prenatal alcohol use.²³ Participants were randomly assigned to the intervention group or a control group. Both the control group and the brief intervention group decreased their use of alcohol after enrolling in the study and undergoing the initial detailed assessment.

For women who were heavier drinkers, the brief interventions for prenatal alcohol use were statistically more effective in reducing their frequency of alcohol consumption, vs the initial assessment alone. In addition, the effects of the brief intervention were significantly enhanced when a support partner of the woman's choice also participated.²³

The Project Choices Intervention Research Group studied an intervention that included 4 sessions of motivational interviewing regarding alcohol habits, and a contraception counseling session.²⁴ Study participants were recruited from 6 community-based settings with high proportions of women at risk for an alcohol-exposed pregnancy, including a jail and 2 drug and alcohol treatment centers. Among the 143 women who completed the 6-month follow-up, 68.5% were no longer at risk of having an alcohol-exposed pregnancy.

These participants successfully lowered their risk by reducing alcohol use only (12.9%), adopting appropriate contraception use only (23.1%), or by changing both risk factors (32.9%). Even if all the study participants who were lost to follow-up were assumed to have been unsuccessful at eliminating their risk of alcohol-exposed pregnancy, more than half of the women (51.6%) successfully changed.²⁴

Take advantage of opportunities

FAS is the most severe consequence of alcohol-exposed pregnancy, leaving the affected child with a lifelong disability. As a family physician, you have access to easy-to-use, cost-effective clinical tools to screen for at-risk drinking behaviors and have sufficient rapport with your patients to encourage effective contraceptive practices. You also have effective tools for helping patients reduce their alcohol consumption.

Within the context of your long-term relationships with patients, you can provide brief interventions that include factual information and opportunities for goal setting. You can assist families with an FAS child to access services, manage medically related complications, and plan for special education and vocational skills training.

Recognition that 1 child in a family is affected by prenatal alcohol exposure gives you another window of opportunity to address the underlying substance use issues in the mother and the family, increasing the odds that future pregnancies will not be alcohol exposed.

JFP

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